EDITORIALS

Birth Defects and Disabilities: A Public Health Issue for the 21st Century

The 20th century witnessed great advances in the diagnosis, treatment, and prevention of birth defects and developmental disabilities and in the quality of life and life expectancy in people living with disabilities. Through newborn screening, early recognition and treatment of phenylketonuria and other metabolic disorders has led to the prevention of mental retardation.[1] The development of new surgical techniques and clinical management of selected birth defects, such as congenital heart disease, spina bifida, and Down syndrome, has resulted in marked increases in survival of children and adults with these conditions.[2,3] Similarly, people living with disabilities have experienced improvements in life expectancy; in the first half of the 20th century, life expectancy for a person with a spinal cord injury was about 14 months; by the end of the century, that rate rose to 46.2 years.[4]

The Children's Health Act of 2000[5] recognized the relevance of those public health issues with the creation of the National Center on Birth Defects and Developmental Disabilities (NCBDDD) at the Centers for Disease Control and Prevention (CDC). That act effectively raised the visibility and scope of successful programs developed since the 1970s at the CDC that were devoted to the prevention of birth defects and developmental disabilities and the promotion of health and wellness in those people living with disabilities. This issue of the Journal is an important step toward recognizing the accomplishments and future challenges of this emerging public health field.

THE SCOPE OF THE PROBLEM

Birth defects are defined as abnormalities of structure, function, or metabolism that are present at birth and result in physical or mental disability or are fatal. About 3% of children born in the United States have a major birth defect; birth defects account for about 20% of all infant deaths.[6] Lifetime costs have been estimated at $6 billion for those infants born in a single year with 1 or more of 17 major birth defects.[7] Some birth defects, such as spina bifida, can be prevented with the periconceptional consumption of folic acid. Subsequent management of infants with spina bifida can reduce medical complications, such as urinary dysfunction and hydrocephalus, and has allowed such individuals to live more independent and productive lives.

Developmental disabilities result in social, emotional, behavioral, cognitive, and motor impairments and include conditions such as cerebral palsy, mental retardation, sensory impairments, autism, and attention deficit hyperactivity disorder. About 17% of children in the United States have a developmental disability, with about 2% having a disability severe enough to require life-long care and special services.[8] Lifetime economic costs of 4 developmental disabilities (mental retardation, cerebral palsy, hearing loss, and vision impairment) were estimated in excess of $50 billion based in 2003 dollars.[9] Ensuring safe and optimal physical and emotional development of all children regardless of the underlying disease, disability, or circumstance is a high priority. We know that early intervention has an important positive impact on children with developmental delays or disabilities.[10]

Disability is defined as an activity limitation associated with a long-term physical, sensory, or cognitive impairment. The US Bureau of the Census estimated that there were approximately 54 million Americans with a disability in 2000, with direct medical costs estimated at $260 billion per year.[11–13] By 2010, because of an aging population, an estimated 70 million Americans will have a disability.[14] The CDC has shown that people who have a disability are more likely to have higher rates of alcohol and tobacco use, engage in less physical activity, be overweight or obese, and use preventive health services, such as breast and cervical cancer screening, less often.[15–16]

Comprehensive public health approaches should focus on ensuring that prevention measures are in place to promote healthy fetal developmental and to prevent the occurrence of birth defects and developmental disabilities. We must also focus on preventing secondary conditions and promoting the health of individuals living with a birth defect, developmental disability, or a disability because of a chronic condition or injury.

ANSWERING THE PUBLIC HEALTH CHALLENGE

To answer the public health challenge, NCBDDD recently completed a long-range strategic planning process, with input from government, nongovernment, and academic partners. We identified 5 areas on which our center can focus so that research and programs will have a greater impact on health The five areas are: (1) promote healthy birth outcomes; (2) ensure optimal child development through early identification and intervention; (3) prevent secondary conditions and promote the health of adolescents and adults living with a disability; (4) identify and eliminate health disparities in certain target groups; and (5) ensure a healthy environment and appropriate emergency response for those in need. These areas taken as a whole reflect a life course approach in dealing with priority health outcomes.

Ensuring healthy birth outcomes includes a strong commitment to public health research to identify the causes of birth defects and developmental disabilities and to use cutting edge science, such as the advances in genomics, in this pursuit. Two major epidemiological research networks have been created with funding from NCBDDD: the National Birth Defects Prevention Study, a collaborative of 9 research sites,[17] and the Centers for Autism and Other Developmental Disabilities Research and Epidemiology, a 6-site collaboration.

Although long-term research investments will advance our understanding of the preventable causes of birth defects and developmental disabilities, these research networks are also designed to be responsive to the emerging public health concerns. For example, public concerns were raised by a recent study in Sweden that suggested a widely prescribed allergy medication, loratadine, was associated with hypospadias, a birth defect of the male genitalia.[18] In response, an analysis of the National Birth Defects Prevention Study data found no association, thus providing important guidance to healthcare providers and pregnant women about this popular medication.[19,20]

One way to ensure healthy birth outcomes is to help women and their partners become knowledgeable about and consider all the appropriate actions that should be taken before and during pregnancy. Messages about proper nutrition; avoiding alcohol, tobacco, and some medications; ensuring adequate exercise; and properly managing chronic medical conditions should be combined into a comprehensive preconception strategy for healthy pregnancies. NCBDDD is taking part in an agency-wide initiative that will explore approaches to bundle prevention messages and to identify barriers to offering preconceptional services.

Carefully monitoring early infant and child development, particularly social and emotional development, is important for ensuring early diagnosis and intervention for developmental problems, such as autism. NCBDDD recently launched a national campaign, "Learn the Signs. Act Early," to educate parents and health care providers about the importance of acting early if there are indications that a child is failing to meet the important and predictable developmental milestones.[21] NCBDDD is also supporting research to examine the feasibility of incorporating a standardized developmental screening protocol into pediatric care. The article by Pinto-Martin et al. in this issue of the Journal discusses challenges in implementing such a program.[22]

Promoting health and well-being in adolescents and adults living with a disability is the third focus area of NCBDDD. As noted by Baker at al. in this issue of the Journal, the development of a comprehensive surveillance and treatment program for hemophilia has led to vast improvement in survival and quality of life in individuals with this condition.[23] Because individuals with a

Photo courtesy of the American Association of AdaptedSports Programs. Photo by Sarah Nash.

Members of Team Holyfield playing in Atlanta, Georgia, in January 2005. Team Holyfield is a member of the National Wheelchair Basketball Association's youth division and is comprised of the most talented male and female athletes from the interscholastic teams of the American Association of AdaptedSports Programs, which sponsors the team in partnership with the Evander Holyfield Foundation. The team shoots at standard 10-foot baskets and competes in regional and national tournaments and conferences as well as with the men's NCAA basketball teams.

chronic condition or disability may not receive the usual prevention messages (i.e., healthy nutrition and physical activity), NCBDDD is educating physicians and other health care providers about the particular needs of people with disabilities and chronic conditions. Caregiving has only recently been considered a public health topic. As Talley and Crews argue in their manuscript (R.C. Talley, PhD, MPH, and J.E. Crews, DPA, unpublished commentary, 2004), to promote optimal health of the recipient, the physical and emotional health of the caregiver must be considered.

To resolve health disparities, the focus by NCBDDD is on special populations: people with disabilities, people living in poverty, and those of racial and ethnic minority groups. A current example is our effort to promote folic acid use in Hispanic women, who have the highest rates of babies with neural tube defects, yet have the lowest daily consumption of folic acid.[24] A targeted, paid media campaign is already showing promise in increasing use of folic acid among this population.

The final area of focus is maintaining a healthy environment and developing emergency procedures for the populations we serve, in particular, pregnant women and people with disabilities. A healthy environment is one that is accessible to all people, and, as Rimmer et al. demonstrate in their article in this issue of the Journal about accessibility in health clubs, there are many challenges to making this vision a reality.[25] NCBDDD is committed to working with CDC colleagues to develop appropriate emergency treatment guidelines for pregnant women to ensure their safety while protecting the well-being of the fetus.[26]

CONCLUSIONS

Birth defects, developmental disabilities, and promoting wellness among people with disabilities are major public health issues that require prompt attention from researchers, public health officials, and the health care system. The causes of birth defects and developmental disabilities must be found. Prevention of injuries and chronic conditions that lead to disabilities must be addressed. Ensuring optimal health for all people with disabilities is a challenge for the health care and public health system that must be acted upon today. ■

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